Your Guide to Understanding Genetic Conditions

CDC6 gene

cell division cycle 6

Normal Function

The *CDC6* gene provides instructions for making a protein that is important in the copying of a cell's DNA before the cell divides (a process known as DNA replication). The protein produced from this gene, called cell division cycle 6 or CDC6, is one of a group of proteins known as the pre-replication complex. In a multi-step process, the components of this complex attach (bind) to certain regions of DNA known as origins of replication (or origins), where the process of DNA copying begins. When the pre-replication complex is attached to the origin, replication is able to begin at that location. This tightly controlled process, called replication licensing, helps ensure that DNA replication occurs only once per cell division and is required for cells to divide.

Health Conditions Related to Genetic Changes

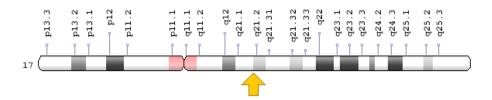
Meier-Gorlin syndrome

At least one mutation in the *CDC6* gene causes Meier-Gorlin syndrome, a condition characterized by short stature, underdeveloped kneecaps, and small ears. This mutation, which is a rare cause of the condition, changes a single protein building block (amino acid) in the CDC6 protein, replacing the amino acid threonine at position 323 with the amino acid arginine (written as Thr323Arg). As a result, assembly of the pre-replication complex is impaired, which disrupts replication licensing; however, it is not clear how a reduction in replication licensing leads to Meier-Gorlin syndrome. Researchers speculate that such a reduction delays the cell division process, which slows growth of the bones and other tissues during development. It is not known why development of the kneecaps and ears is particularly affected in Meier-Gorlin syndrome.

Chromosomal Location

Cytogenetic Location: 17q21.2, which is the long (q) arm of chromosome 17 at position 21.2

Molecular Location: base pairs 40,287,689 to 40,303,161 on chromosome 17 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- CDC6 cell division cycle 6 homolog
- CDC6-related protein
- CDC6_HUMAN
- cdc18-related protein
- CDC18L
- cell division control protein 6 homolog
- cell division cycle 6 homolog
- HsCDC6
- HsCDC18
- p62(cdc6)

Additional Information & Resources

Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): DNA Synthesis Begins at Replication Origins https://www.ncbi.nlm.nih.gov/books/NBK26826/#_A796_
- - https://www.ncbi.nlm.nih.gov/books/NBK9940/# A789

Scientific Articles on PubMed

PubMed

https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28CDC6%5BTIAB%5D%29+OR+%28cell+division+cycle+6%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D

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Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology http://atlasgeneticsoncology.org/Genes/CDC6ID40014ch17q21.html
- ClinVar https://www.ncbi.nlm.nih.gov/clinvar?term=CDC6%5Bgene%5D
- HGNC Gene Symbol Report http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/ hgnc_data.php&hgnc_id=1744
- NCBI Gene https://www.ncbi.nlm.nih.gov/gene/990
- UniProt http://www.uniprot.org/uniprot/Q99741

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